Tools for Polyploids Workshop
Computational Support

SNP and Dosage Calling

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TOOLS FOR POLYPLOIDS

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Getting Prepared for the Workshop

- Polyploids
- Molecular Markers
- Genome variations – applications
  - Quantitative traits mapping
  - Genome Wide Association studies
  - Phenotypic predictions - Genome Selection
  - Evolution and diversity studies
  - Gene expression studies

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Genome variations

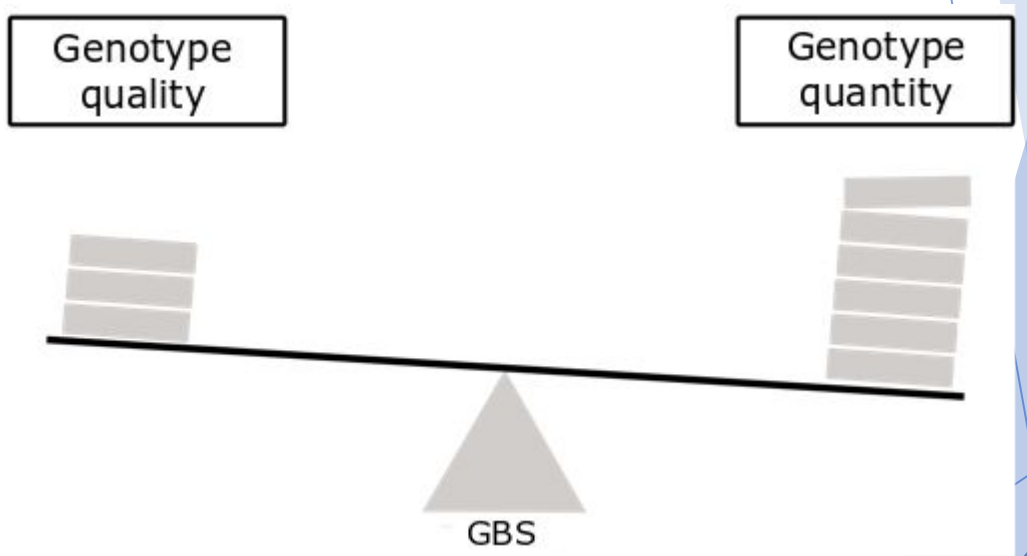
- Short sequences (SNPs, indels)
- Structural variants (number of copies, inversions, translocations)

Molecular markers

- RFLP, RAPD, AFLP, and SSR
- Arrays (For Roses: $$$$$)
- Sequencing (For Roses: $)

Sequencing Experiment Design

- Study goal
- Sequencer capacity
- Number of individuals per lane
- Number of sequenced loci



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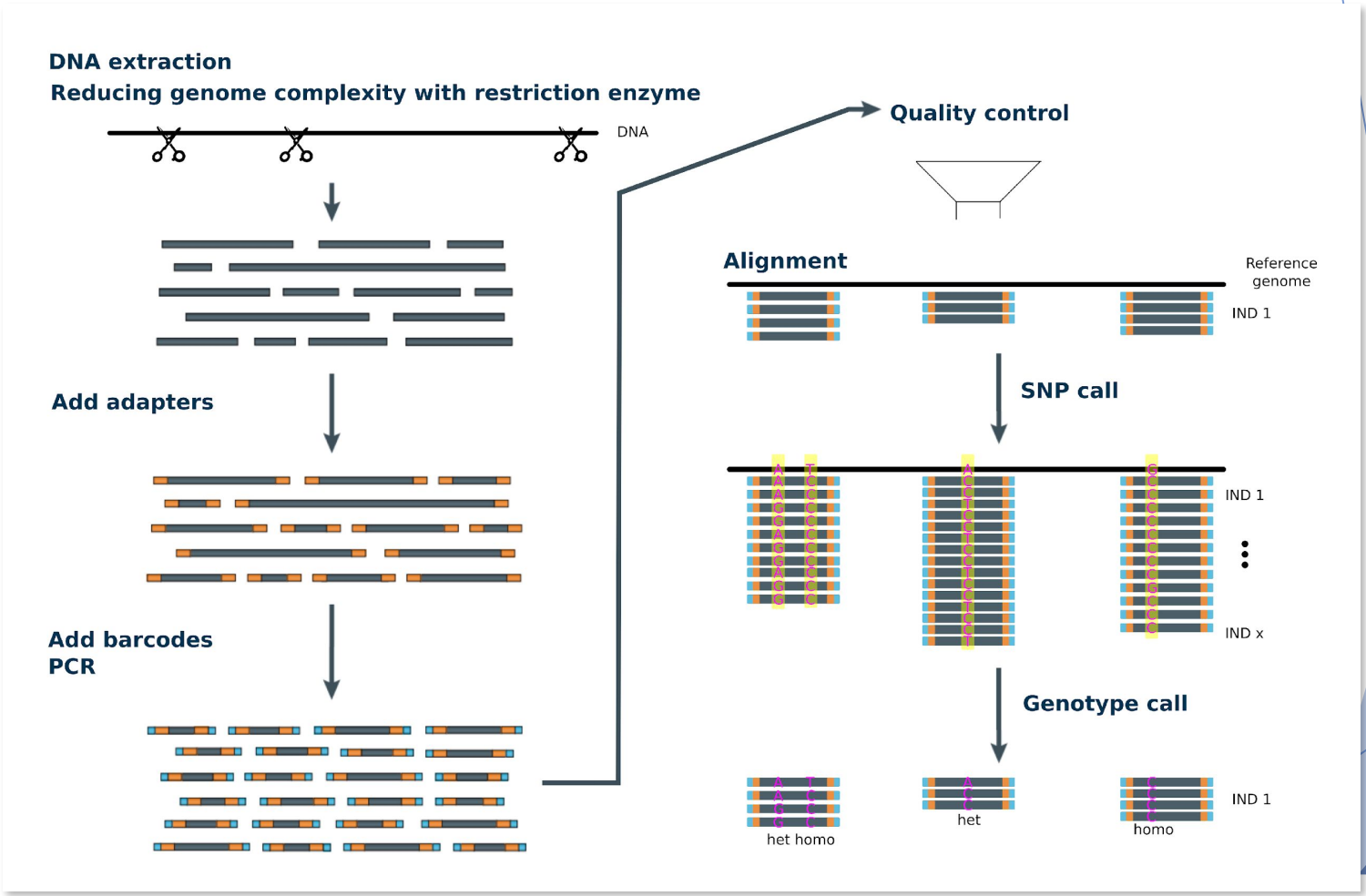
GBS Overview



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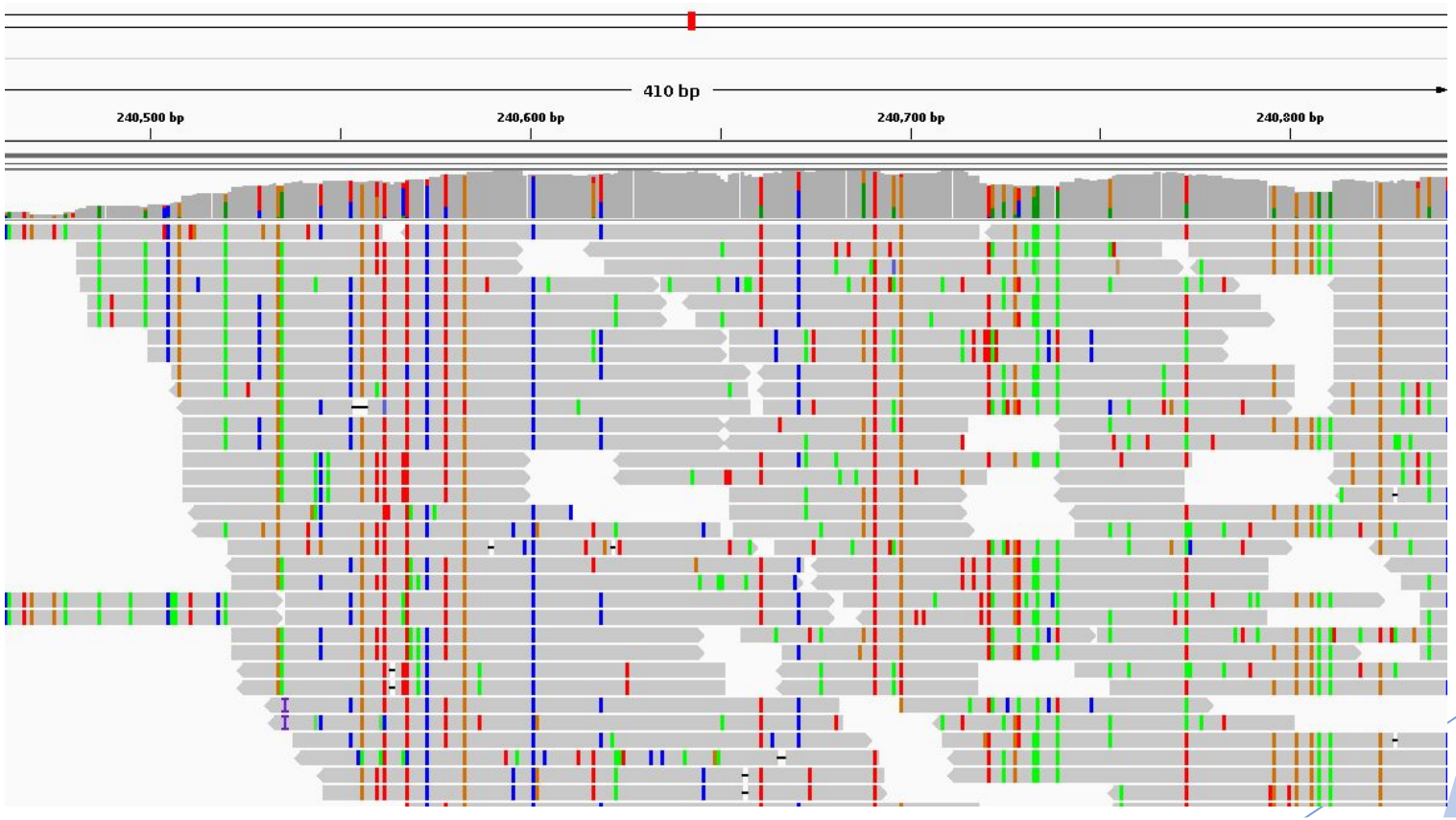
SNP Calling

- Whole Genome Sequencing (WGS)

Image: IGV



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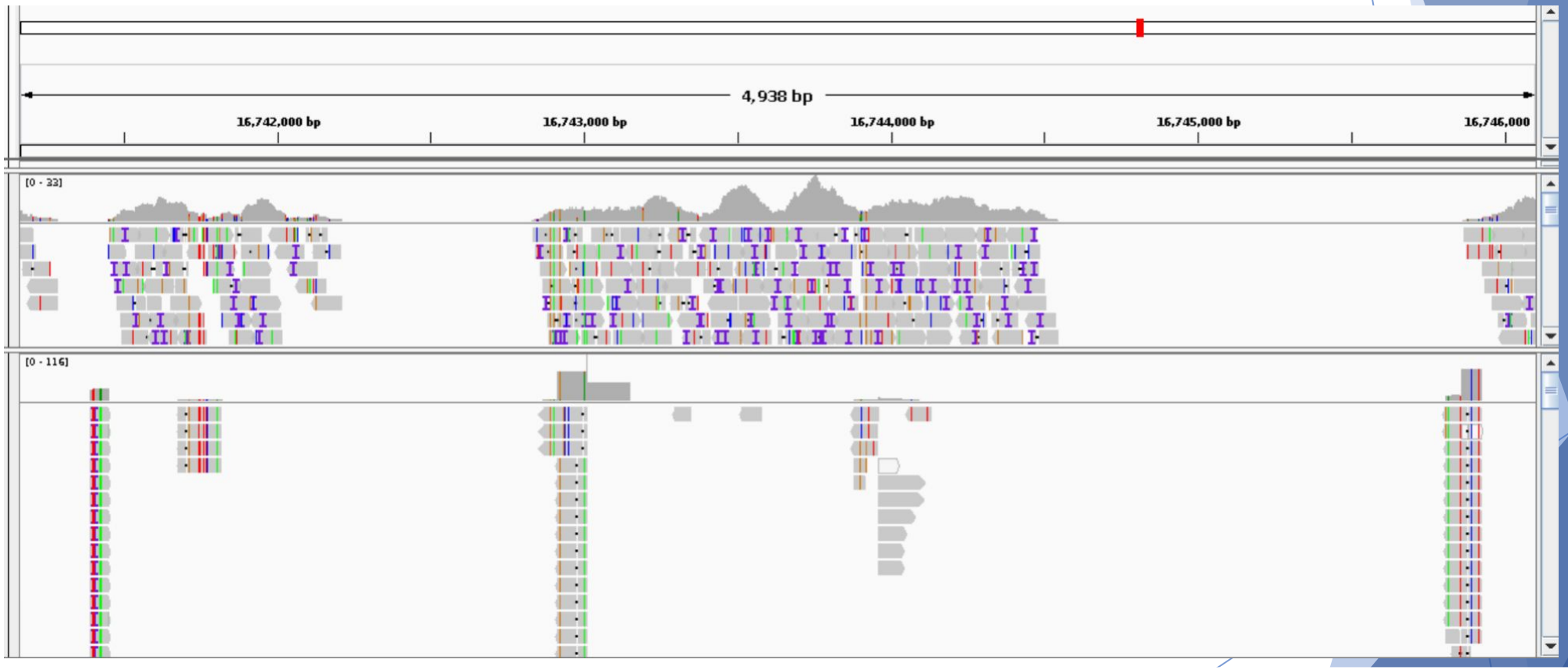
SNP Calling

- Exome sequencing (top) and Genotyping-by-Sequencing (bottom)

Image: IGV



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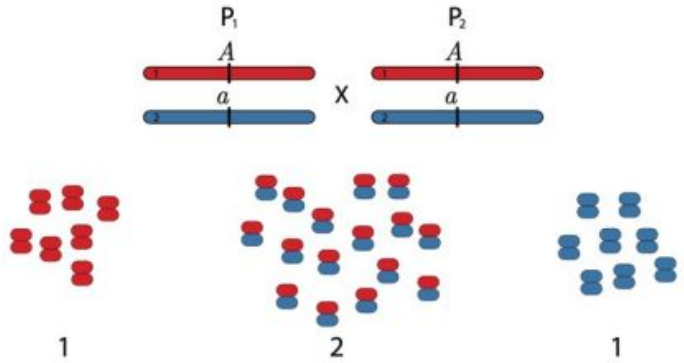
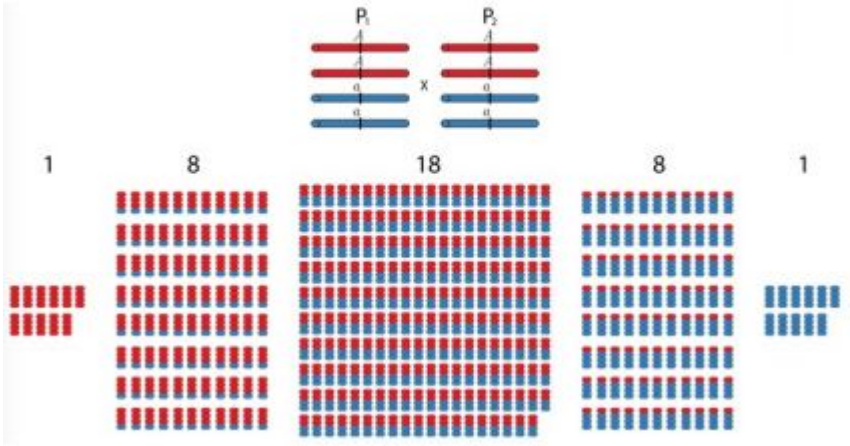
Dosage calling

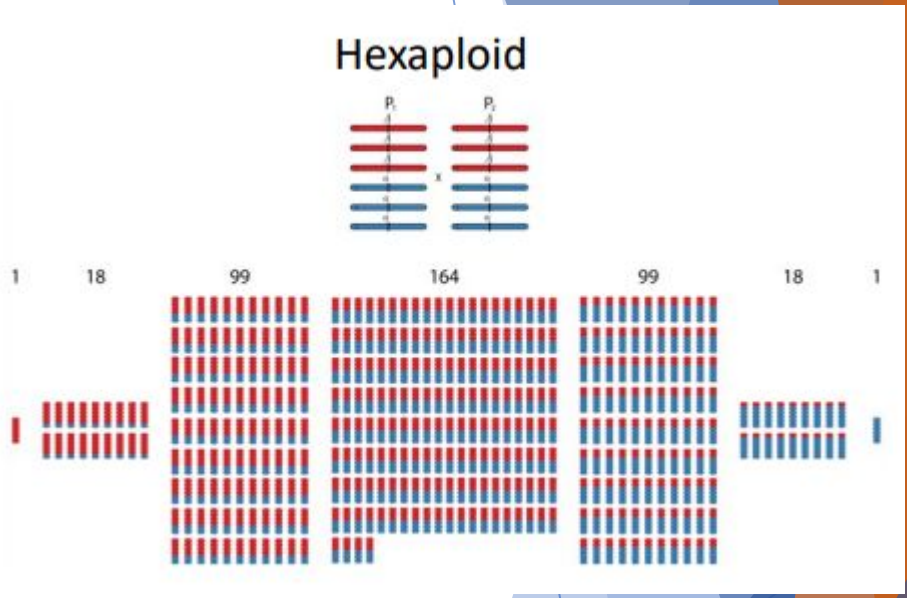
Diploid



Tetraploid



Hexaploid



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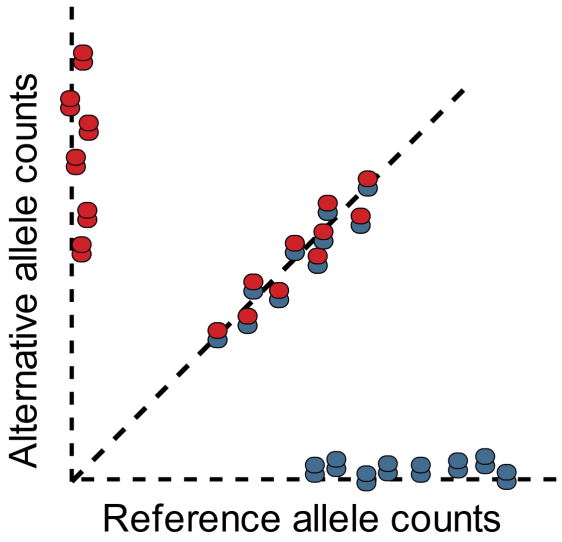
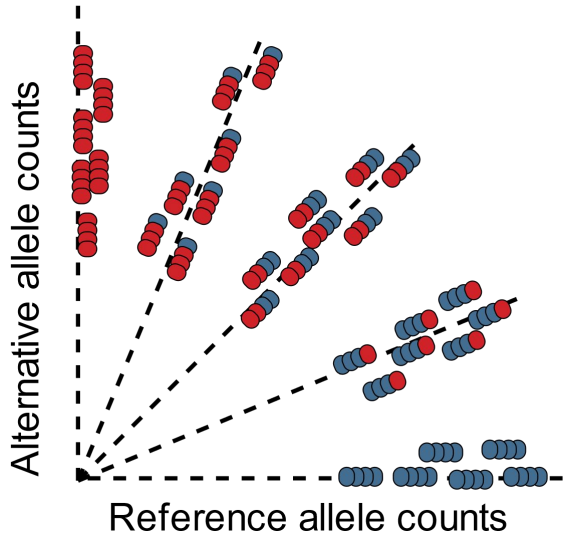
Dosage Calling

- The theory

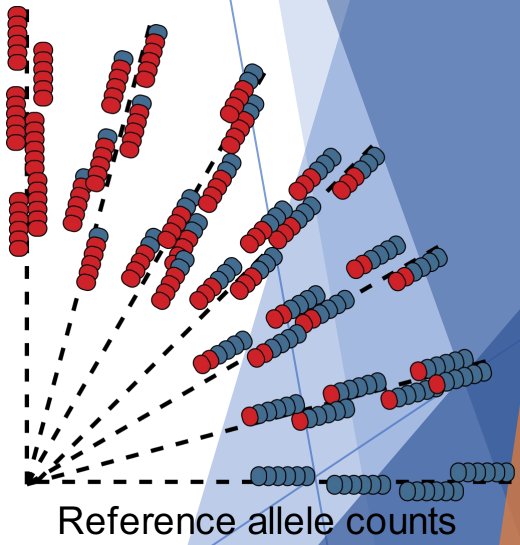
Diploid



Tetraploid



Hexaploid



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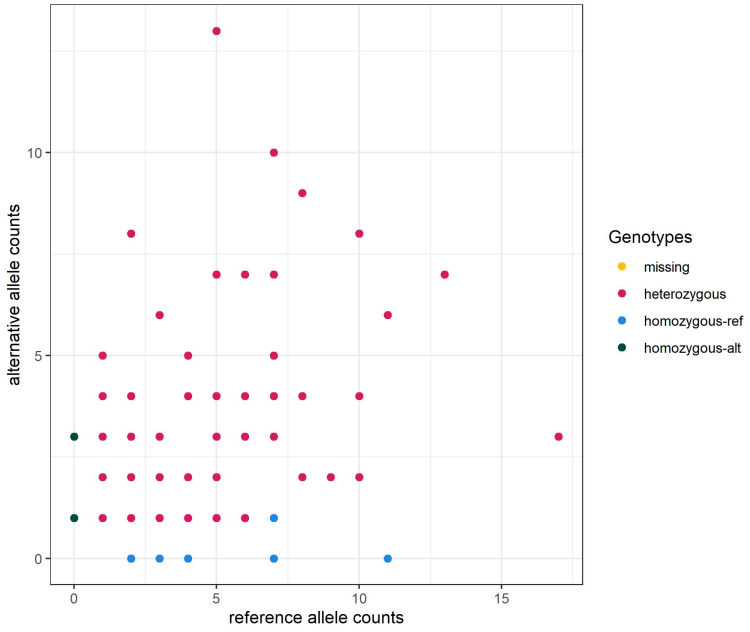
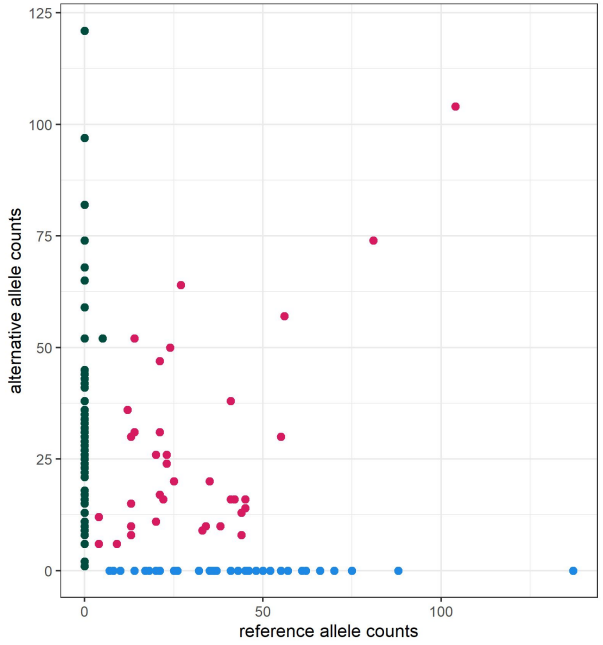
Dosage Calling

- The reality

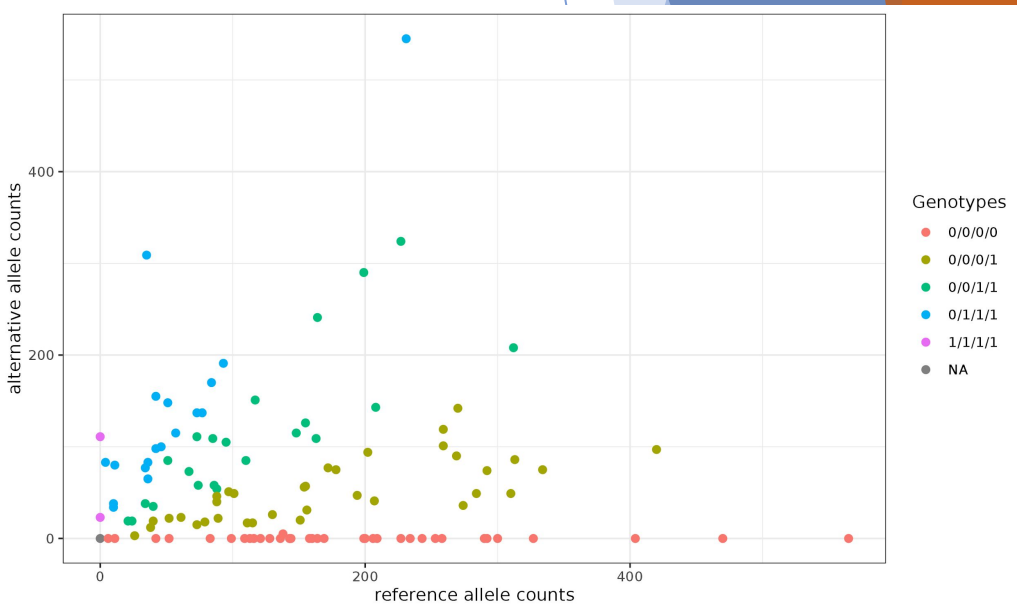
Diploid (mean depth 6)
N = 200
Aa x Aa



Diploid (mean depth 96)
N = 138
Aa x Aa



Tetraploid (mean depth 83)
N = 114
AAaa x AAaa



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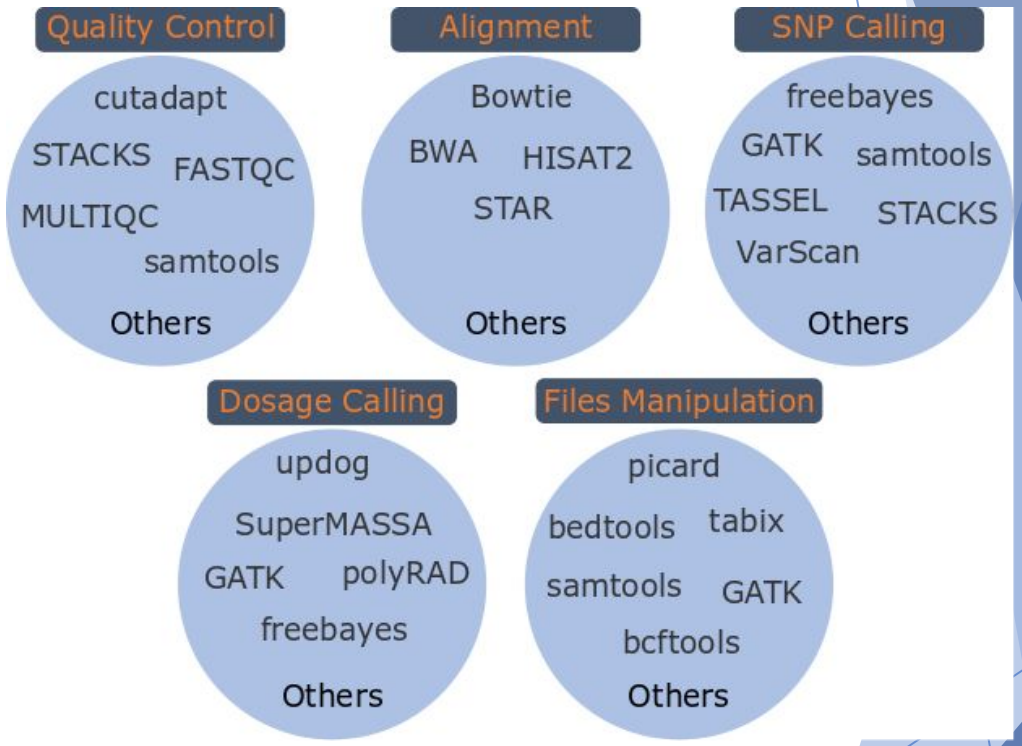
Sequencing Data - Technical Difficulties

- Large files

- Many software
- Many programming languages
- Different Operational Systems
- Updates



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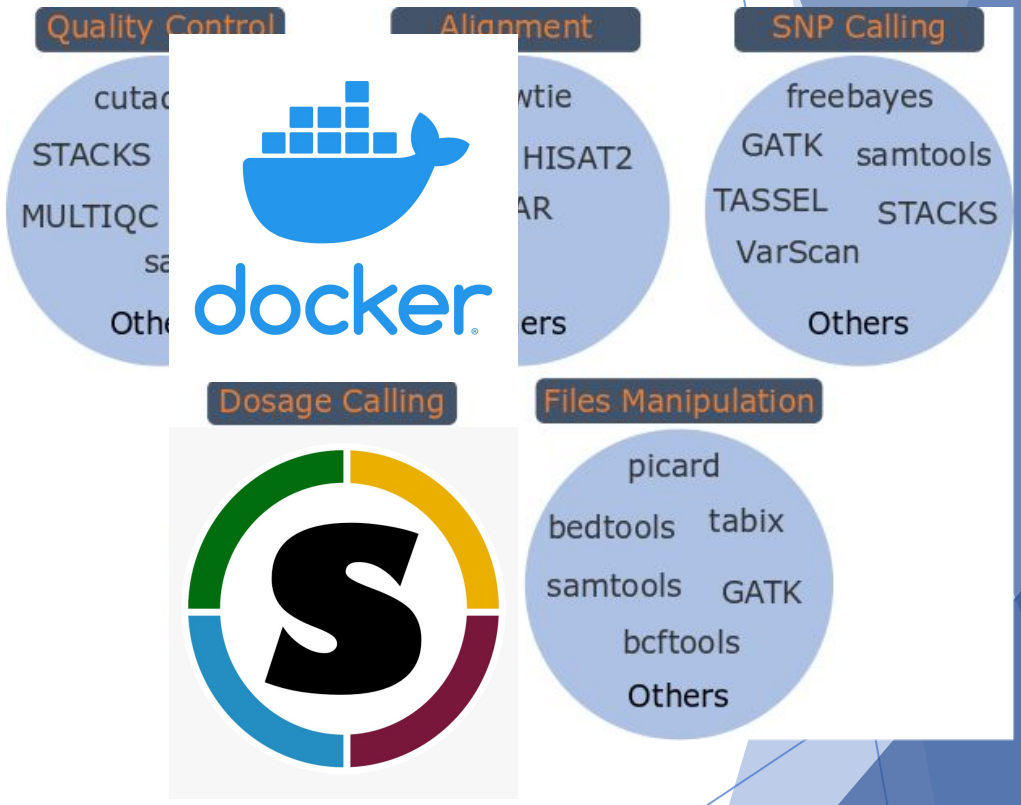
Sequencing Data - Technical Difficulties

- Large files
  - High Performance Computing (HPC)
  - Management systems (SLURM, SGE)
  - Cloud (Google, Amazon)

- Many software
- Many programming languages
- Different Operational Systems
- Updates
  - Containers
    - Docker
    - Singularity (usually available in HPC)
    - BioContainers



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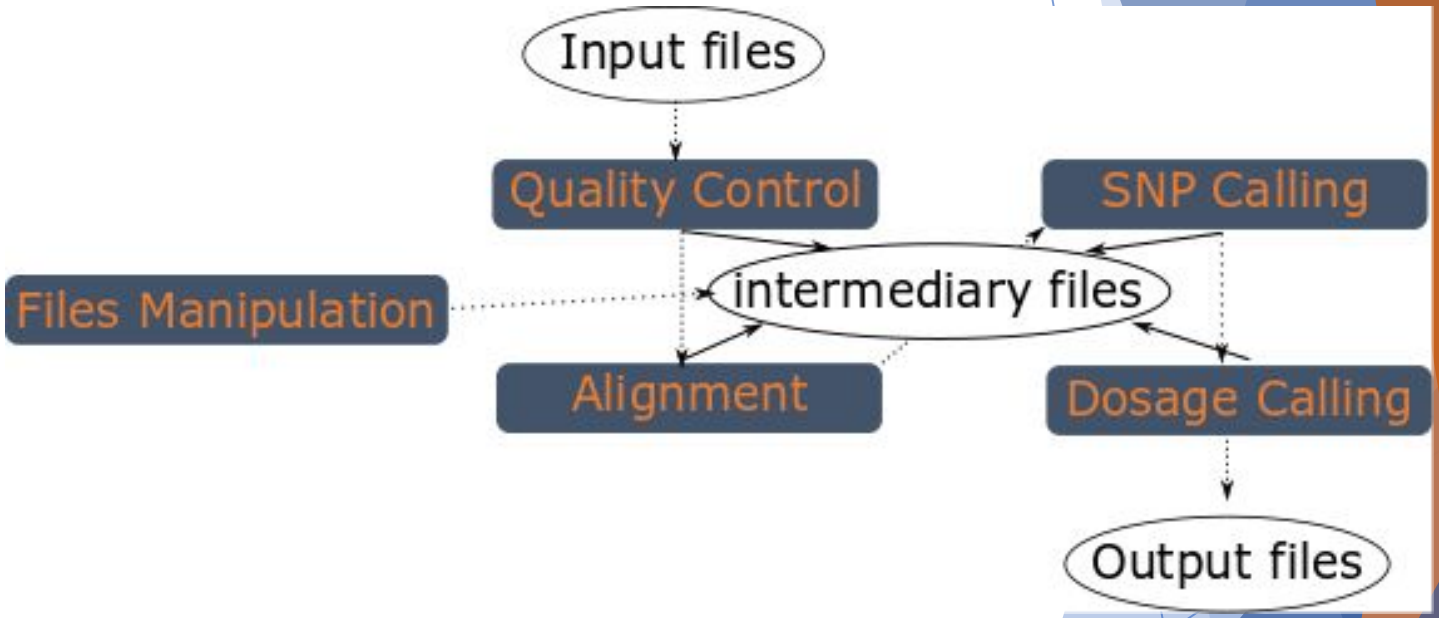
Sequencing Data - Technical Difficulties

- Many steps
- Many file formats



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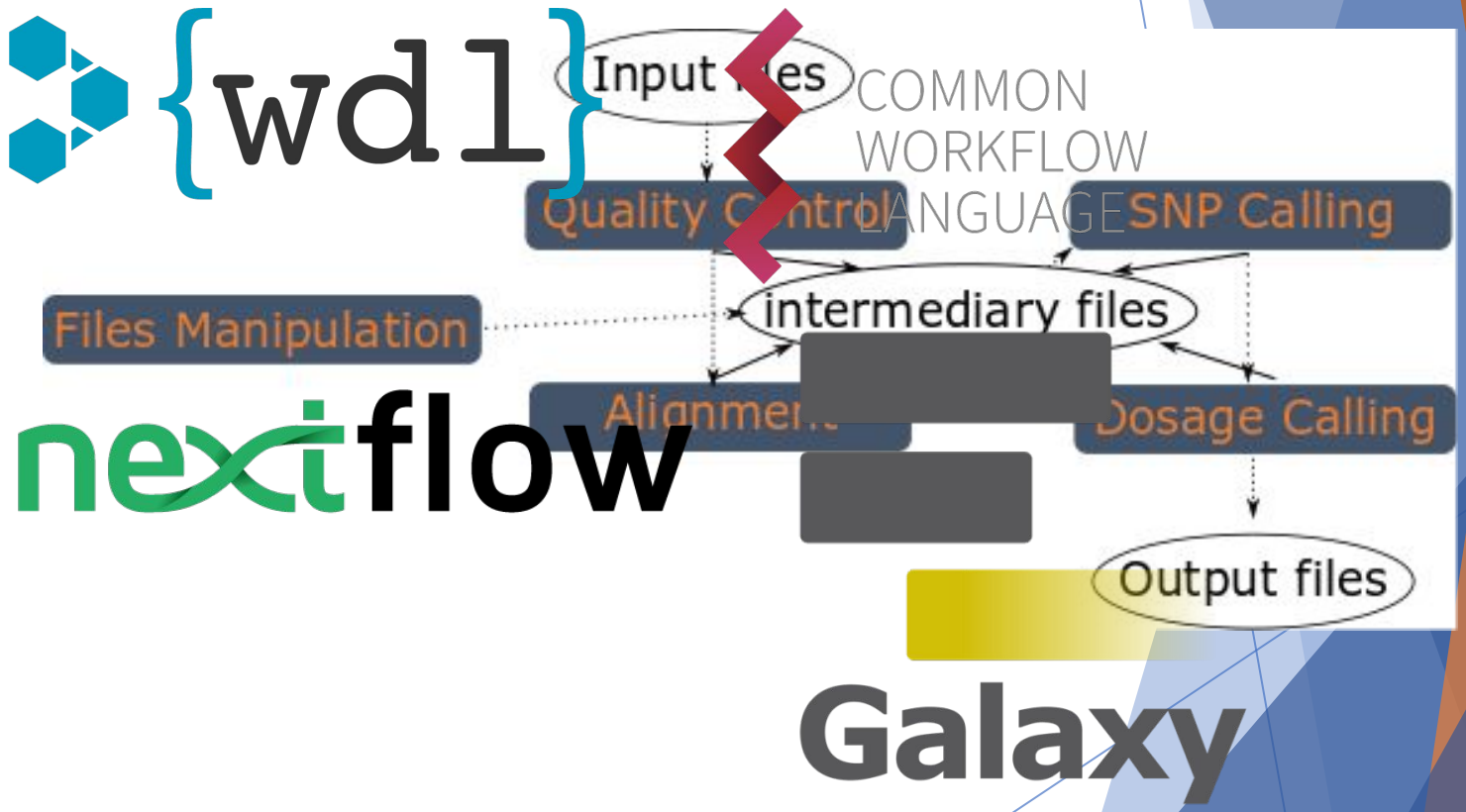
Sequencing Data - Technical Difficulties

- Many steps
- Many file formats
  - Workflows systems
    - Galaxy
    - Nextflow
    - Snakemake
    - CWL
    - WDL
  - Workflows repositories
    - Dockerstore
    - WorkflowHub
  - Run workflows on Cloud
    - Galaxy
    - DNAnexus
    - Terra
    - AnVIL
    - SevenBridges



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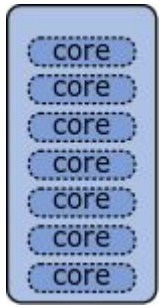
Sequencing Data - Technical Difficulties

- Resources optimization
  - Time
  - Cores
  - Nodes
  - RAM memory

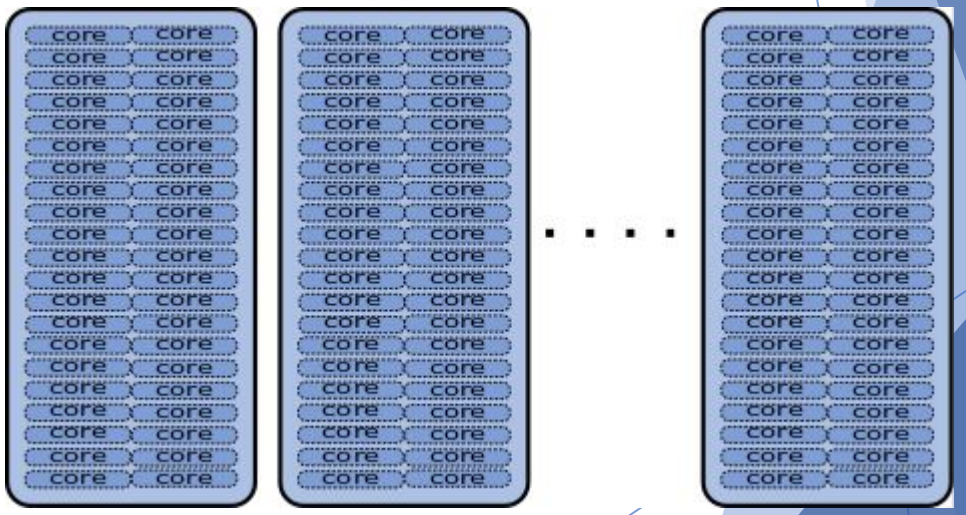
Personal Computer:

4GB RAM; 8 cores; 1 node



High Performance Computing (Texas A&M):

384GB; 48 cores per node; 900 nodes



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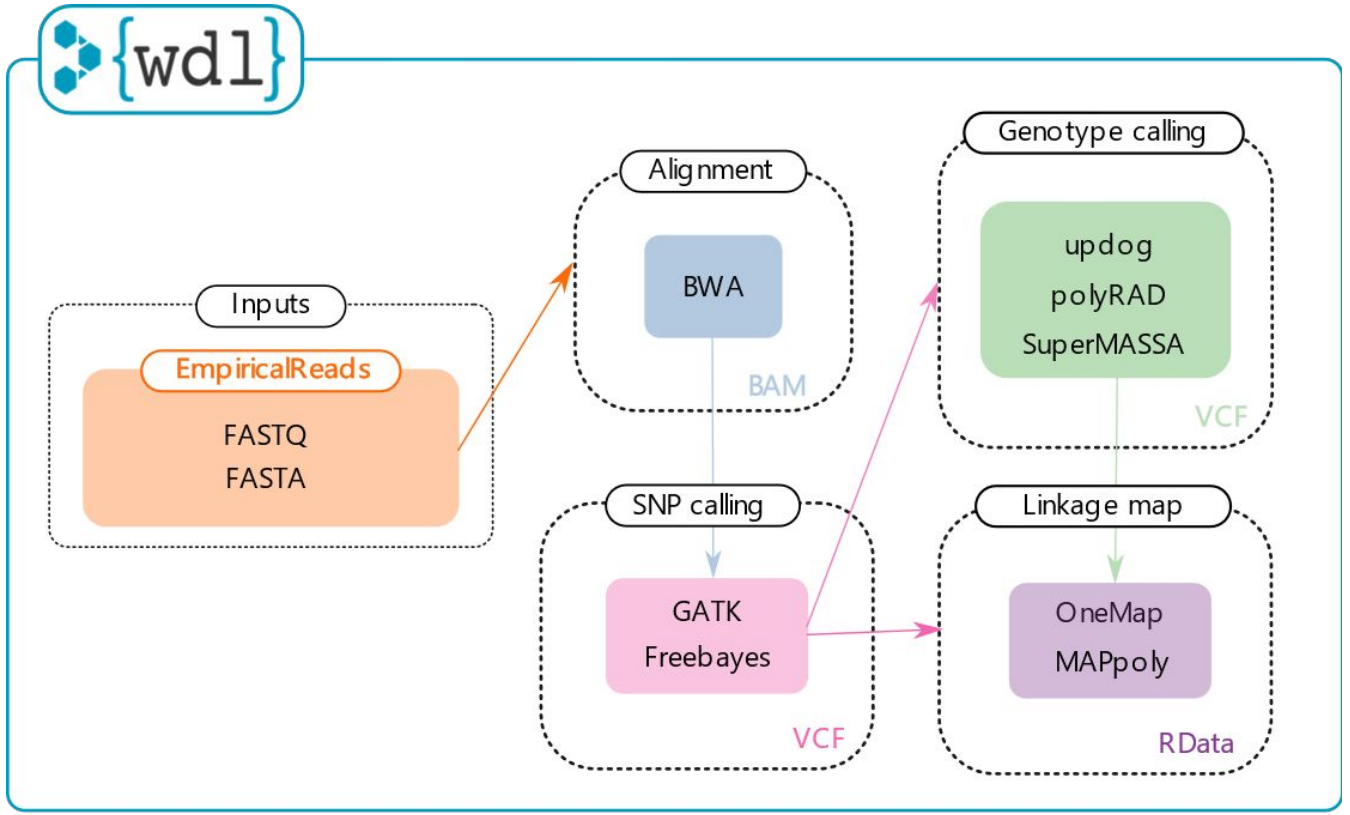
Reads2Map



Available in Github, Dockerstore and WorkflowHub

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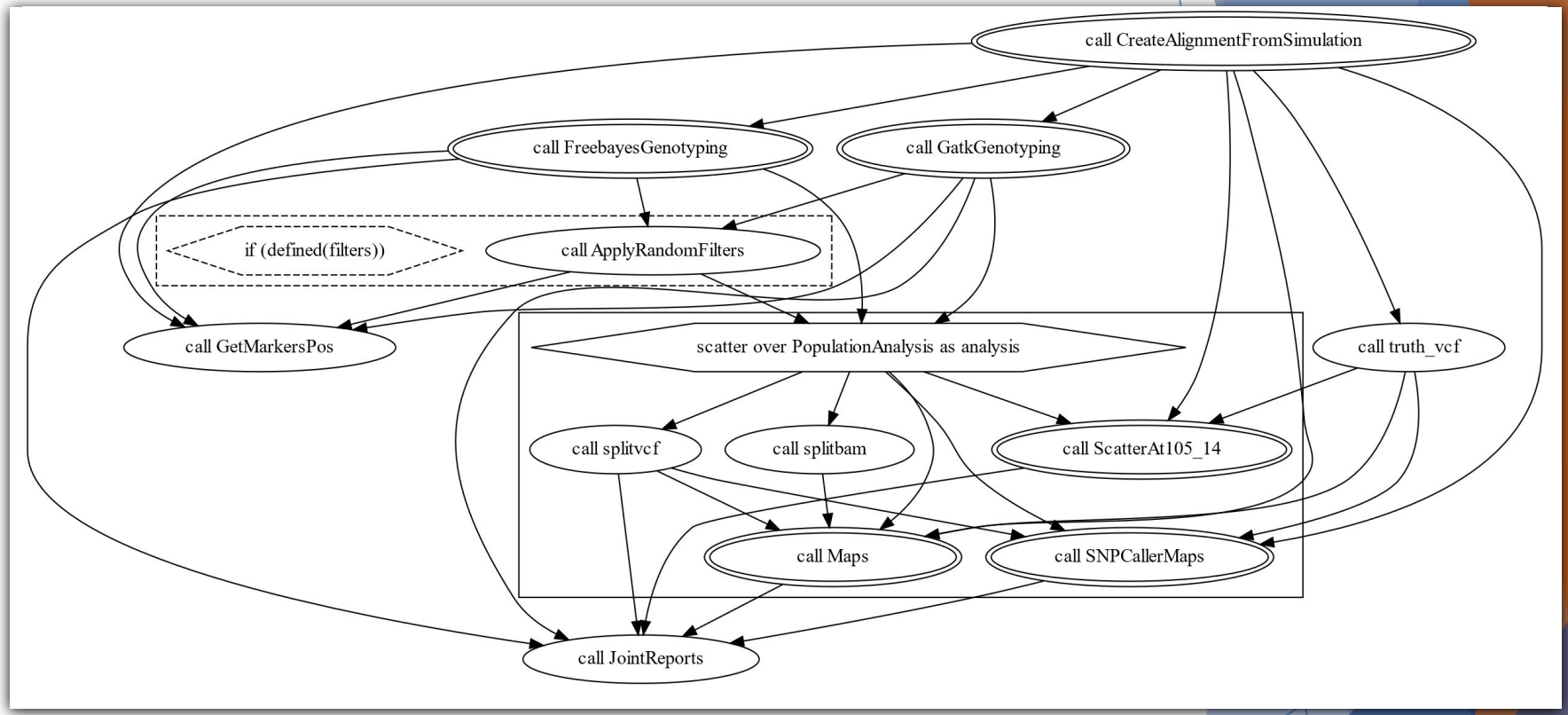
Implementation

- Workflows
  - Sub-workflows
    - Tasks




```
$ java -jar /path/to/womtool.jar graph tasks/SimulatedSingleFamily.wdl > SimulatedSingleFamily.dot
$ dot –Tsvg SimulatedSingleFamily.dot –o SimulatedSingleFamily.svg
```

TOOLS FOR POLYPLOIDS

Implementation

- Cloud environments
  - terra.bio
- High Performance Computing (HPC)
  - Cromwell
  - MiniWDL
  - dxWDL

```
$ java -jar /path/to/cromwell.jar run -i inputs/EmpiricalSNPCalling.inputs.json
EmpiricalSNPCalling.wdl
```

Tutorials

- polyRAD tutorial
- updog tutorial
- fitPoly tutorial
- (TASSEL) Variant and Genotype Calling in Highly Duplicated Genomes (Lindsay Clark)
- Step-by-step of SNP and dosage calling using containers and WDL workflows

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References

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